TruGenome Technical Sequence Data

Immediate access to high-quality, clinical-grade whole-genome sequencing.

Some clinical laboratories have the expertise to analyze and interpret whole-genome sequencing data, but are not set up to perform the actual sequencing. With the TruGenome Technical Sequence Service, you can take advantage of whole-genome sequencing in the CLIA-certified, CAP-accredited Illumina Clinical Services Laboratory and perform your own clinical analyses and interpretation for your patients.

What is the TruGenome Technical Data Service?
The TruGenome Technical Service uses the industry-leading Illumina next-generation sequencing (NGS) technology to provide accurate clinical whole-genome sequencing. Filters for calling bases have been established based on extensive validation experiments, and are set to ensure analytical sensitivity of > 98% and analytical specificity of > 99%. The test provides broad coverage of exomic, promoter, and regulatory regions. Variants are annotated to facilitate interpretation, although complete interpretation is not included.

What information do I receive from this service?
The TruGenome Technical Service delivers a report outlining the methodology used and specifications of the sequencing run and an encrypted hard drive containing sequencing data with all reads, quality scores, and variants for further analysis. BAM files contain aligned and unaligned reads of the genome. gVCF and VCF files include the nucleotide calls and the quality of each call made at each position that passed filter throughout the genome. If needed, support from the Illumina team of genetic specialists is available. A complete description of the TruGenome Technical Sequence Service is available at www.illumina.com/test_descriptions.

What is involved in using the Technical Service?
The first step is to determine if you have a need for whole-genome sequencing and access to the bioinformatics capabilities and clinical expertise necessary for analyzing and interpreting the data. Next, request a Sample Collection Kit from Illumina. Use the provided forms to obtain patient consent and the collection tubes to procure a blood sample from your patient. Send the completed paperwork and blood sample to the Illumina Clinical Services Laboratory for processing. Technical data will be provided to you on a hard drive for your analysis and interpretation.

Using the TruGenome Technical Sequence Data Service*

Contract with Illumina to perform whole-genome sequencing.

Collect patient's blood sample and send to Illumina.

Sample received and sequencing performed in the Illumina Clinical Service Laboratory.

Hard drive containing technical data in the form of BAM, gVCF, and VCF files sent to customer.

* This graphic provides a high-level overview of the process for using the Technical Service from Illumina. For complete details, visit www.illumina.com/clinicallab.

About the Illumina Clinical Services Laboratory

The CLIA-certified, CAP-accredited Illumina Clinical Services Laboratory was established in 2009 for the purposes of offering human whole-genome sequencing services to physicians. It is staffed with certified Clinical Laboratory Scientists who have extensive training and expertise with Illumina NGS technology. Our team of Ph.D. geneticists and bioinformaticians, genetic counselors, and certified medical geneticists has over 50 years of combined experience analyzing human genetic data.

About Illumina NGS technology

Illumina sequencing by synthesis (SBS) technology is the most published NGS technology. Using a proprietary method, DNA sequence is detected in a base-by-base manner for highly accurate, reproducible sequencing results. Learn more about sequencing at www.illumina.com/sequencing.

Learn more

If you are considering whole-genome sequencing for your patients, we strongly encourage you to contact us before ordering so that we can help you choose the appropriate test and aid in diagnostic evaluation. We are available to discuss coverage of the targeted genes/regions, additional analysis and/or support that might be needed, and any other testing needs. To learn more about the Illumina Clinical Services Laboratory and the Technical Service, visit www.illumina.com/clinicallab.

Ordering information

Service	Turnaround Time (TAT)	Catalog No.
TruGenome Technical Sequence Service	Standard (45 days)	FT-800-1001
	Rapid (14 days)†	FT-800-1011

† The rapid turnaround test must be arranged in advance to make sure that a system is reserved to sequence the samples upon arrival.